



DNAI1 gene

dynein axonemal intermediate chain 1

Normal Function

The *DNAI1* gene provides instructions for making a protein that is part of a group (complex) of proteins called dynein. This complex functions within cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells. Coordinated back and forth movement of cilia can move the cell or the fluid surrounding the cell. Dynein produces the force needed for cilia to move.

Within the core of cilia (the axoneme), dynein complexes are part of structures known as inner dynein arms (IDAs) and outer dynein arms (ODAs) depending on their location. Coordinated movement of the dynein arms causes the entire axoneme to bend back and forth. IDAs and ODAs have different combinations of protein components (subunits) that are classified by weight as heavy, intermediate, or light chains. The *DNAI1* gene provides instructions for making intermediate chain 1, which is found in ODAs. Other subunits are produced from different genes.

Health Conditions Related to Genetic Changes

heterotaxy syndrome

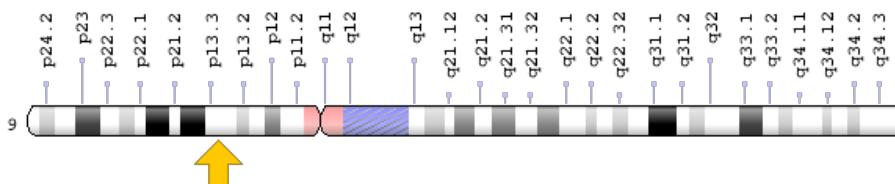
primary ciliary dyskinesia

At least 21 mutations in the *DNAI1* gene have been found to cause primary ciliary dyskinesia, which is a condition characterized by respiratory tract infections, abnormal organ placement, and an inability to have children (infertility). *DNAI1* gene mutations result in an absent or abnormal intermediate chain 1. Without a normal version of this subunit, the ODAs cannot form properly and may be shortened or absent. As a result, cilia cannot produce the force needed to bend back and forth. Defective cilia lead to the features of primary ciliary dyskinesia.

Chromosomal Location

Cytogenetic Location: 9p13.3, which is the short (p) arm of chromosome 9 at position 13.3

Molecular Location: base pairs 34,458,752 to 34,520,989 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- axonemal dynein intermediate chain 1
- CILD1
- DIC1
- DNAI1_HUMAN
- dynein intermediate chain 1, axonemal
- dynein intermediate chain DNAI1
- dynein, axonemal, intermediate chain 1
- dynein, axonemal, intermediate polypeptide 1
- IC78
- ICS1
- immotile cilia syndrome 1
- MGC26204

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Cilia and Flagella: Structure and Movement
<https://www.ncbi.nlm.nih.gov/books/NBK21698/>

GeneReviews

- Primary Ciliary Dyskinesia
<https://www.ncbi.nlm.nih.gov/books/NBK1122>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28DNAI1%5BTIAB%5D%29+AND+%28primary+ciliary+dyskinesia%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- DYNEIN, AXONEMAL, INTERMEDIATE CHAIN 1
<http://omim.org/entry/604366>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DNAI1%5Bgene%5D>
- HGNC Gene Family: Dyneins, axonemal
<http://www.genenames.org/cgi-bin/genefamilies/set/536>
- HGNC Gene Family: WD repeat domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/362>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2954
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/27019>
- UniProt
<http://www.uniprot.org/uniprot/Q9UI46>

Sources for This Summary

- OMIM: DYNEIN, AXONEMAL, INTERMEDIATE CHAIN 1
<http://omim.org/entry/604366>
- Escudier E, Duquesnoy P, Papon JF, Amselem S. Ciliary defects and genetics of primary ciliary dyskinesia. *Paediatr Respir Rev.* 2009 Jun;10(2):51-4. doi: 10.1016/j.prrv.2009.02.001. Epub 2009 Apr 18. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19410201>
- Failly M, Saitta A, Muñoz A, Falconnet E, Rossier C, Santamaria F, de Santi MM, Lazor R, DeLozier-Blanchet CD, Bartoloni L, Blouin JL. DNAI1 mutations explain only 2% of primary ciliary dykinesia. *Respiration.* 2008;76(2):198-204. doi: 10.1159/000128567. Epub 2008 Apr 23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18434704>

- GeneReview: Primary Ciliary Dyskinesia
<https://www.ncbi.nlm.nih.gov/books/NBK1122>
- Leigh MW, Pittman JE, Carson JL, Ferkol TW, Dell SD, Davis SD, Knowles MR, Zariwala MA. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet Med.* 2009 Jul;11(7):473-87. doi: 10.1097/GIM.0b013e3181a53562. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19606528>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3739704/>
- Pennarun G, Escudier E, Chapelin C, Bridoux AM, Cacheux V, Roger G, Clément A, Goossens M, Amselem S, Duriez B. Loss-of-function mutations in a human gene related to Chlamydomonas reinhardtii dynein IC78 result in primary ciliary dyskinesia. *Am J Hum Genet.* 1999 Dec;65(6):1508-19.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10577904>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1288361/>
- Zariwala MA, Leigh MW, Ceppa F, Kennedy MP, Noone PG, Carson JL, Hazucha MJ, Lori A, Horvath J, Olbrich H, Loges NT, Bridoux AM, Pennarun G, Duriez B, Escudier E, Mitchison HM, Chodhari R, Chung EM, Morgan LC, de Jongh RU, Rutland J, Pradal U, Omran H, Amselem S, Knowles MR. Mutations of DNAI1 in primary ciliary dyskinesia: evidence of founder effect in a common mutation. *Am J Respir Crit Care Med.* 2006 Oct 15;174(8):858-66. Epub 2006 Jul 20.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16858015>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2648054/>
- Zietkiewicz E, Nitka B, Voelkel K, Skrzypczak U, Bukowy Z, Rutkiewicz E, Huminska K, Przystalowska H, Pogorzelski A, Witt M. Population specificity of the DNAI1 gene mutation spectrum in primary ciliary dyskinesia (PCD). *Respir Res.* 2010 Dec 8;11:174. doi: 10.1186/1465-9921-11-174.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21143860>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3014902/>

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